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NEUROLOGY CONSULTATION

[redacted]

JULY 1, 2003

[redacted] is a 26-year-old woman I was asked by [redacted] to see in neurological consultation for bilateral proximal leg weakness.

[redacted] was a very active varsity athlete in high school and rowed crew at [redacted] for her first year of college before giving it up to concentrate on studies. Somewhere around her junior and senior year of college she noticed slight weakness in her legs but felt it was because she was not working out as much as before. This has slowly progressed over the intervening years, and she finally came to the realization that it was not just because she was not working out. Her exercise tolerance was less, and even when she worked out more, she did not improve. Therefore, a few months ago she saw [redacted] for an electrodiagnostic study at Sturdy Memorial Hospital, but before that study was completed, he requested that [redacted] take over, presumably because of her prior training in neuromuscular diseases. [redacted] subsequently did a very thorough nerve conduction and EMG study on May 9, 2003, which showed normal sensory and motor nerve conduction studies of the arms and legs but chronic neurogenic changes in the proximal leg muscles bilaterally. [redacted] subsequently saw [redacted] in consultation and performed additional testing, including an MRI of the lumbosacral spine and a lumbar puncture. Because of concerns about motor neuron disease, and in particular spinal muscular atrophy, [redacted] asked me to see her in consultation.

[redacted] denies any problems with her arms. She denies any problems with swallowing or chewing. There has been no change in her speech. She has myopia but no other change in her vision. She has no problem with bowel or bladder function. She denies any pain. She does note that she has difficulty going up stairs and often has to use the rail. She also cannot get out of a chair without using her hands. If she is tired, she will begin to feel as if she is losing her balance. She does not know if she can run because she has not tried. She has not noticed any decrease in the size of her legs. She denies any sensory loss.

PAST MEDICAL HISTORY: Significant for attention deficit disorder and learning disability diagnosed during high school. She has been on Ritalin since then and continues on 15 mg. t.i.d. She recently had an evaluation in Boston by a neuropsychologist, the results of which are unknown to me. An MRI of her brain was suggested which she recently had at Sturdy Memorial Hospital, and I do not have that report either.

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FAMILY HISTORY: Positive for multiple sclerosis on her mother's side in an uncle and a cousin. Her mother has chronic Lyme disease and, in fact, several other family members have also had Lyme disease (she is from Old Lyme, Connecticut). Her father died in 1997 of myelodysplasia.

SOCIAL HISTORY: She went to [redacted] for high school and completed four years of college at [redacted], graduating with a Bachelor of Fine Arts. She does not smoke. She uses alcohol only socially. She currently lives with two roommates and works as a fabric designer in [redacted]

REVIEW OF SYSTEMS: The review is documented on the history form she filled out and is positive for some weight change, joint pain, and weakness.

EXAMINATION: She is awake, alert, oriented x 4, fluent, and cooperative. She looks her stated age and is in no acute distress. She was in mild emotional distress as she gave me the history, crying on one occasion and being teary-eyed on another. Her pulse and respiratory rate are normal. She has a normal temperature. Cranial nerves II-XII are completely intact. Her fundus examination is normal. Motor examination shows good strength of all of her neck muscles, as well as her deltoids, biceps, triceps, wrist extensors, finger extensors, and interossei bilaterally. In her lower extremities, her iliopsoas is 4 bilaterally, quadriceps 5, and her hamstrings are also 5. Her tibialis anterior, tibialis posterior, peroneus longus, gastrocnemius, and extensor hallucis longus are also 5 bilaterally. Her adductors are 4 on the right and 4+ on the left. Her gluteus medius is 4 bilaterally. Her gluteus maximus is 5 to 5- bilaterally. Her deep tendon reflexes are 2+ and symmetrical in the biceps, triceps, brachioradialis, knees, and ankles. She has normal tone in all four limbs. She has full range of motion in all joints. Her plantar responses are downgoing bilaterally. Her sensory examination is intact to touch, pin, and vibration in all four limbs. Her finger-nose-finger, gait, stance, and Romberg are normal. I detect no pelvic waddle when she walks. She has no Trendelenburg sign when she lifts one leg. I find no scoliosis. She is able to do one sit-up easily. She cannot rise from a chair without using her hands. She cannot do a deep knee-bend. She can climb up onto a stool easily with one leg.

DATA: MRI of the cervical spine showed minimal levoscoliosis with its apex at C7 and mild degenerative change. MRI of the lumbar spine was normal except for a 6 mm. extraspinal synovial cyst adjacent to the lateral aspect of the right facet joint at L4-5. Spinal fluid examination showed 0 white blood cells, a protein of 26, and a glucose of 57. The cerebrospinal fluid had a normal RPR, cryptococcal antigen, and Lyme titer. Other laboratory work showed a CPK of 222, which is minimally elevated. She had a sedimentation rate of 6. She had a normal vitamin B12, folate, C-reactive protein, and rheumatoid factor.

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ADDENDUM: I just received the report of her MRI of the brain which showed global cerebellar atrophy and a small area of FLAIR signal abnormality next to the right temporal horn without enhancement.

IMPRESSION: [redacted] has mild bilateral proximal leg weakness which appears to be indolently progressive over the past six to seven years. She has no family history. The EMG is suggestive of a neurogenic process, which raises the issue of whether she might have type III spinal muscular atrophy, also known as Kugelberg-Welander disease. Other possibilities include myopathic entities, such as muscular dystrophy. I do not believe she has amyotrophic lateral sclerosis or juvenile amyotrophic lateral sclerosis. The abnormality on the MRI of her brain is interesting, but I suspect it is long-term and unrelated to her progressive proximal leg weakness.

PLAN: I have suggested that we obtain the genetic test for spinal muscular atrophy, which is looking for deletion in the survival motor neuron gene at chromosome 5. If that is positive, it would confirm a diagnosis of spinal muscular atrophy, and I would see her in return for genetic counseling related to that. If it is negative, it does not completely exclude that diagnosis, but it would then require a muscle biopsy to further pursue. I will arrange that appointment once the results are back.

[redacted]

cc: [redacted]

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